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A case of Noonan's syndrome in a 6-year-old boy is presented in which xanthomas of the skin and tongue were an additional, not previously reported, finding. The other syndromal stigmata are essentially those reported by Noonan and others in the literature. This entity has only rarely been discussed in the dental literature although a great number have been reported in the medical literature.

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Noonan's Syndrome; Report of a Case with Oral Findings

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In 1928 Turner[1] described seven female patients whose dominant presenting clinical signs were sexual infantilism, congenital webbed neck and cubitus valgus. Subsequently it was noted that female patients such as those he described had only 45 chromosomes with an XO karyotype.[2] Phenotypically male patients have also been presented with the superfical stigmata of "Turner's syndrome" and were referred to as "Turner's syndrome in the male."[3] Confusion reigned as how this seemingly incongruous situation could occur with chromosomal studies indicating, in most cases, a normal chromosomal configuration with a diploid number of 46 and no recognizable abnormalities.

In 1963 Noonan and Ehmke[4] described nine male and female patients who presented with similar facies, cardiac abnormalities and multiple extracardiac anomalies including small stature, hypertelorism, ptosis, cryptorchidism, short and sometimes webbed neck, low-set ears and a slight antimongoloid slant to the eyes. They indicated that no evidence was present indicating chromosomal abnormality in the syndrome. Noonan[5] further elaborated on these and additional cases and on the basis of her findings the entity has become widely known as "Noonan's syndrome." Familial patterns have been subsequently demonstrated.[6,7,8]

Recent workers[6-7] have concluded that Noonan's syndrome is a separate entity

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from Turner's syndrome and occurs almost exclusively in normal karyotypes. It may, however, be transmitted as an autosomal dominant trait with incomplete penetrance thus accounting for the familial patterns and isolated sporadic cases.

Congenital cardiac defects are not an uncommon entity nor is it unusual for these defects to be a part of a syndrome complex. Many syndromes also present with oral and maxillo-facial manifestations of which abnormal jaw relations and/or cleft palates are the dominating features.

The purpose of this paper is to present a case of Noonan's syndrome in a six-year-old male. In addition to the common systemic features associated with this condition, pertinent oral features are illustrated. A high arched palate and hypoplastic mandible have been previousely described.[3,8] However, the additional finding of multiple dermatological and lingual xanthomas is, to the authors' knowledge, the first such report in the dental literature.

Case Report

The patient, a six-year-old caucasian male, was referred to the dental service at Walter Reed Army Medical Center for an oral evaluation prior to cardiac surgery for the correction of a stenotic pulmonary valve. The oral examination revealed an intact dentition in a good state of repair with no carious lesions present clinically or on X-ray. The patient had a high, narrow palatal vault and multiple small white nodules on the dorsum of the tongue (Fig. 1). The latter has been present since birth as determined from the history. The physical examination revealed a small but well-developed caucasion male with curly hair and a peculiar facies (Fig. 2). He wore glasses and his conjunctivae were inflammed with multiple petechia present. The right eye displayed exotropia and both eyes exhibited epicanthal folds. The ears were low-set, large and were scaley and erythematous. The neck was short although not webbed. The skin was dry with multiple pustules, nodules and scars over the abdomen, extremities and face.

The chest was shield-shaped with widely spread nipples and lateral flailing of the lower costal margins. He had severe edema below the knees. There were no testes in the scrotum and the patient had phimosis of the penis. Previous extensive workups had indicated a fused urinary collection system with only one ureter entering the bladder and with a possible ureterocele. The patient had a grade III/IV systolic ejection murmur, first degree AV block and right ventricular hypertrophy. A catheterization procedure had revealed pulmonic valve stenosis and an atrial septal defect. Biopsy procedures of the skin lesions resulted in the diagnosis of xanthoma (Fig. 3). Laboratory procedures performed prior to cardiac surgery including blood studies, electrolytes and an SMA-12/60 were all within normal limits. Chest X-rays and an electrocardiogram confirmed the cardiac abnormalities. The patient had no siblings and his natural parents displayed none of the abnormalities noted in the patient.

On June 5, 1975, the patient underwent suture repair of the atrial septal defect and resection of the dysplastic tricuspid pulmonary valve under total cardiopulmonary bypass. He did reasonably well post-operatively but on June 13 the patient manifested a left plural effusion. This increased in severity necessitating multiple drainage procedures. On July 12, after apparent overall improvement, the patient underwent cardiopulmonary arrest and died.

Discussion

Congenital heart disease was present in all of Noonan's original and subsequent cases. Valvular pulmonary stenosis was the dominant specific cardiac abnormality although atrial septal defects and a patent ductus arteriosus were also noted. The present case demonstrated both pulmonary stenosis and a septal defect each of which was felt to be of a severe enough nature to necessitate the high-risk surgery which ultimately resulted in the patient's demise. The bilateral and longstanding edema of the lower extremities has been reported[3] and is presumably

secondary to the compromised cardiac status.

Urogenital defects including cryptorchidism and various developmental abnormalities of the kidney and ureters have been shown to be a part of Noonan's syndrome and were noted in the present case.

Skeletal defects in addition to the basically small stature occur primarily in the thoracic area. Abnormal nipple placement and/or configuration have presented as features of the syndrome and were noted in this case.

Antimongoloid slant of the eyes, low set ears and short neck have been consistant findings of the syndrome and all were evident in this patient. Noonan discussed hypertelorism as an important entity in her cases but it's presence was questionable in this instance.

This case of Noonan's syndrome is felt to be of interest as an addition to the literature primarily due to the previously unreported occurrence of multiple skin and oral xanthomas. These abnormalities, while not debilitating or life-threatening in themselves, are of interest from a diagnostic standpoint. Many syndromes such as Gardner's syndrome, basal cell nevoid syndrome and others present with various types of dermatologic lesions as well as lesions of the oral mucosa and within the jaws. Cleft palate or abnormal palatal configuration is a perplexing problem and is one of the most commonly occuring intraoral features of many syndromes. A high arched palate has been established as a feature of Noonan's syndrome and was seen in the present case. Xanthomas of the skin and tongue had not, however, been previously reported and while their presence may be fortuitous, it appears likely that these lesions are a vital part of the syndrome complex. It will be of interest to note if future reports substantiate or repudiate this finding.

Summary

A case of Noonan's syndrome in a 6-year-old boy is presented in which xanthomas of the skin and tongue were an additional, not previously reported, finding. The other syndromal stigmata are essentially those reported by Noonan and others in the literature. This entity has only rarely been discussed in the dental literature although a great number have been reported in the medical literature.

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Illustrations

Fig. #1 Dorsum of the tongue with multiple nodules.

Fig. #2 Facial features with antimongoloid slant of eyes, short neck and low-set ears.

Fig. #3 Biopsy of nodule illustrating xanthoma cells (H&E Stain Magnification X250)